

20177YP

TITLE OF THE INVENTION
BEST'S MACULAR DYSTROPHY GENE

ABSTRACT OF THE DISCLOSURE

- 5 Novel human and mouse DNA sequences that encode the gene
CG1CE, which, when mutated, is responsible for Best's macular dystrophy, are
provided. Provided are genomic CG1CE DNA as well as cDNA that encodes the
CG1CE protein. Also provided is CG1CE protein encoded by the novel DNA
sequences. Methods of expressing CG1CE protein in recombinant systems are
10 provided. Also provided are diagnostic methods that detect patients having mutant
CG1CE genes.